Disease	Cause (Deficiency of)	Symptoms and manifestations	Treatment
Steatorrhea	Deficiency of pancreatic lipase: In case of chronic pancreatitis or pancreatectomy ➔indigestion of fat. Diseases of the liver: hepatitis or biliary sys disease (chronic cholycystitis or bile duct obstruction) ➔ deficiency of bile salts ➔ no fat solubilization ➔ color of stool may be very light. Diseases in intestinal mucosa ➔ decrease absorption of lipids.	Presence of fat in stool ➔ milky feces or fatty diarrhea.	
Chyluria (Milky Urine)	Congenital abnormal connection (fistula) between lymphatic drainage of the intestine (thoracic duct) and the urinary system.	Presence of chylomicrons in urine after a fatty meal.	The fistula must be removed surgically.
Metabolic disorders of FA β-oxidation	Deficiency of carnitine, carnitine-palmetoyl transferase I or II (CPT I, CPK II) & acyl-CoA dehydrogenase.	Impaired FA β-oxidation lead to: - Fasting hypoglycemia. - Muscle weakness. - Fatty liver. - Coma and death (finally).	
Zellweger's syndrome (Cerebro-hepato-renal syndrome)	Inherited absence of peroxisomes in all tissues.	Accumulation of long chain FA in brain, liver and kidney. Usually die early at age (6 months).	Can be diagnosed prenatally by examination of amniotic fluid for long chain FA or peroxisomal enzymes.
Refsum's disease	Congenital deficiency of enzyme system of α-oxidation.	Accumulation of phytanic acid or its derivatives in nervous system ➔ nervous damage (Deafness, Blindness, Ataxia or Polyneuritis)	
Deficiency of PUFA	Deficiency of Poly Unsaturated Fatty Acids (PUFA).	- Fatty liver in adults. - Decreased normal growth and dermatitis in infants.	
Sphingolipidosis (Lipid storage diseases) — **Gaucher's disease**	Genetic deficiency of lysosomal enzyme: β-Glucosidase.	Accumulation of Glucoceramide.	Accumulation of these complexes especially in neurons ➔ neuro-degeneration, liver enlargement, mental retardation & may be fatal in early life.
Sphingolipidosis (Lipid storage diseases) — **Niemann's disease**	Genetic deficiency of lysosomal enzyme: Sphingomyelinase.	Accumulation of sphingomyelin.	
Multiple sclerosis	Autoimmune disease (antibodies against myelin sheath).	Demyelination of nerves of white matter. There is loss of phospholipids and sphingolipids from white matter. The lipid composition of white matter resembles that of grey matter. The CSF shows high levels of IgG and phospholipids.	
Ketosis	- Prolonged fasting or starvation. - High fat & low carbohydrates in diet. - Severe uncontrolled diabetes mellitus. - Prolonged administration of anti-insulin hormones especially in diabetic persons. - Prolonged and severe muscular exercise.	- Acidosis: if not treated may lead to academia➔ decrease bicarbonate ➔ coma or even death. - Electrolyte loss especially potassium ➔ dehydration.	- Glucose intravenous in case of fasting or starvation. - Glucose & insulin in case of diabetes. - Bicarbonate to correct acidosis. - Potassium in case of hypokalemia (especially in diabetics) & fluids in case of dehydration.

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Disorders of Plasma Lipoproteins

Types	Subtypes		Causes	Symptoms
Hyperlipo-proteinemia	Primary Hyperlipo-proteinemia	Type I: Deficiency of plasma lipoprotein lipase	Deficiency of plasma lipoprotein lipase.	Increase plasma level of chylomicrons and VLDL.
		Type II: Familial hypercholesterolemia	Defect in LDL receptors in liver and other tissues.	Marked increase in LDL.
		Type III: Familial hyperlipoprotienemia	Deficiency of remnants clearance by liver cells.	Increase in chylomicrons & VLDL remnants levels.
		Type IV: Familial hypertriacylglycerolemia	Overproduction of VLDL.	Usually associated with: - Coronary heart diseases. - Type II diabetes mellitus. - Obesity. - Alcoholism.
	Secondary Hyperlipo-proteinemia		Duo to other diseases like: - Diabetes mellitus. - Obstructive jaundice. - Hypothyroidism. - Nephrotic syndrome. - Obesity.	
Hypolipo-proteinemia	A-beta-lipoproteinemia		Failure of synthesis of apo-β, defective formation of chylomicrons, VLDL & LDL.	
	Deficiency of LCAT		Deficiency of LCAT	Marked decrease in cholesterol esters and HDL.

Fatty Liver

Causes	Tests related to lipid metabolism
<u>Over feeding of fats:</u> The uptake of fat by liver cells is more than the capacity to synthesize VLDL.	<u>Cholesterol esters/Total cholesterol ratio</u> Liver is the site for synthesis of LCAT that converts free cholesterol to cholesterylesters. In case of liver diseases, this ratio is decreased.
<u>Over feeding carbohydrates:</u> This stimulates lipogenesis.	
<u>Over mobilization of fat from adipose tissue:</u> As in diabetes mellitus, very low carbohydrates in diet & prolonged starvation.	
<u>Decreased oxidation of fatty acids:</u> This produces increase in free FA available for esterification and TAG synthesis. This occurs in the following conditions: 1- Deficiency if pantothenic acid needed for synthesis of coenzyme A. 2- Deficiency of methyl donors needed for synthesis of carnitine. Excessive intake of nicotinic acid produces depletion of methyl donors as it is excreted in urine as N-methylnicotinamide.	<u>Estimation of plasma bile acids</u> Plasma bile acids level decreased in case of hepatic cell disease.
<u>Deficiency of lipotropics:</u> These are needed for synthesis of lipoproteins ➔ decrease mobilization of fat from liver to extra-hepatic tissues.	
<u>Liver toxins:</u> As CCl_4, chloroform, asenic and phosphorus. They decrease protein synthesis or fatty acid oxidation.	
<u>Alcoholism:</u> Ethyl alcohol is oxidized in liver cells by alcohol dehydrogenase to acetate, which produces increase in fatty acid synthesis and esterification.	

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